SH2D1A gene

SH2 domain containing 1A

Normal Function

The *SH2D1A* gene provides instructions for making a protein called signaling lymphocyte activation molecule (SLAM) associated protein (SAP). SAP interacts with other proteins called SLAM family receptors to activate signaling pathways that are involved in the control of immune cells (lymphocytes). In particular, it helps regulate lymphocytes that destroy other cells (cytotoxic lymphocytes) and is necessary for the development of specialized lymphocytes called natural killer T cells. SAP also helps control immune reactions by triggering self-destruction (apoptosis) of lymphocytes when they are no longer needed.

Health Conditions Related to Genetic Changes

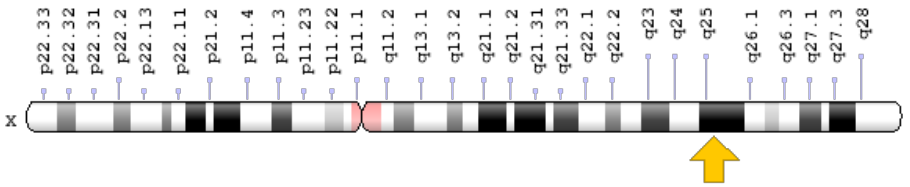
X-linked lymphoproliferative disease

More than 70 *SH2D1A* gene mutations have been identified in people with X-linked lymphoproliferative disease (XLP). Some *SH2D1A* gene mutations impair SAP function. Others result in an abnormally short protein that is unstable or nonfunctional, or prevent any SAP from being produced. The loss of functional SAP disrupts proper control of the immune system and may result in the life-threatening immune reaction to Epstein-Barr virus infection that occurs in this disorder. In addition, cancers of immune system cells (lymphomas) may develop in affected individuals when defective lymphocytes are not properly destroyed by apoptosis.

Chromosomal Location

Cytogenetic Location: Xq25, which is the long (q) arm of the X chromosome at position 25

Molecular Location: base pairs 124,346,282 to 124,373,160 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- DSHP
- Duncan disease SH2-protein
- EBVS
- MTCP1
- SAP
- SH2 domain-containing protein 1A
- SH21A_HUMAN
- signaling lymphocyte activation molecule-associated protein
- SLAM-associated protein
- XLP
- XLPD

Additional Information & Resources

Educational Resources

- Immunobiology (fifth edition, 2001): X-linked lymphoproliferative syndrome is associated with fatal infection by Epstein-Barr virus and with the development of lymphoma
  https://www.ncbi.nlm.nih.gov/books/NBK27109/#A1513

GeneReviews

- Lymphoproliferative Disease, X-Linked
  https://www.ncbi.nlm.nih.gov/books/NBK1406

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28SH2D1A%5BTIAB%5D%29+OR+%28%28LYP%5BTIAB%5D%29+OR+%28SAP%5BTIAB%5D%29+OR+%28XLP%5BTIAB%5D%29+OR+%28DSHP%5BTIAB%5D%29+OR+%28EBVS%5BTIAB%5D%29+OR+%28XLPD%5BTIAB%5D%29+OR+%28MTCP1%5BTIAB%5D%29+OR+%28SLAM-associated+protein%5BTIAB%5D%29+OR+%28signaling+lymphocyte+activation+molecule-associated+protein%5BTIAB%5D%29+OR+%28signaling+lymphocytic+activation+molecule-associated+protein%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D

OMIM

- SH2 DOMAIN PROTEIN 1A
  http://omim.org/entry/300490

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SH2D1A.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SH2D1A%5Bgene%5D
- HGNC Gene Family: SH2 domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/741
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10820
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/4068
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https://ghr.nlm.nih.gov/gene/SH2D1A

Reviewed: August 2010
Published: March 21, 2017

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